Duschenne Muscular Dystrophy

X-LINKED RECESSIVE

3,500 to 6,000 males at birth

1 in 50,000,000 females at birth

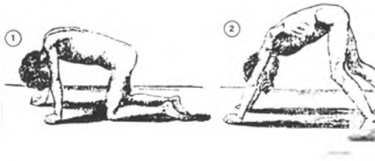
The average life expectancy is 26;

Cause: mutation in the gene for
the protein dystrophin. Dystrophin is important to maintain the muscle fiber's cell membrane.

Muscle weakness usually begins around the age of four, and worsens quickly. Muscle loss typically occurs first in the thighs and pelvis followed by the arms. This can result in trouble standing up. Most are unable to walk by the age of 12. Some may have intellectual disability.

Genetic testing can often make the diagnosis at birth. Those affected also have a high level of creatine kinase in their blood.

No cure, treatment is supportive in natu



Duchenne's Muscular Dystrophy

Sex-linked recessive inheritance

Mother normal, carrier

Father normal

Only males affected, but females may be carriers

♂ 2 yrs old, affected

♂ 5 yrs old, normal

♂ 8 yrs old, affected

♀? 10 yrs old, normal; may or may not be carrier

♂ 15 yrs old, affected

2 years

8 years

15 years

Minimal or no symptoms

Severe crippling deformities and contractures

Progression with age { Weakness, especially of pelvic girdle muscles; marked lordosis, enlarged calves

Calf muscles usually but not always enlarged

Lordosis disappears when child sits